“III European Days of Albinism Milan”

Milan, 7-8 April 2016
Thursday, 7 April 2016
08:00 - 08:30 Registration

08:30-09:00 General welcome
Chair: Tronconi E, Marocchi A, Gesu G, Representative of the Region, Masto R, Paschetta L

09:00-10:00 Scientific Session I
Clinical Definition of Albinism
Chair: Gottlob I, Piozzi E

- Expanding the phenotype of albinism? - Barbara Käsmann-Kellner (20 min)
- Phenotypic spectrum in FHONDA and albinism – Maria Van Genderen (20 min)
- Discussion (20 min)

10:00 - 11.30 Scientific Session II
Clinical and Molecular Diagnosis of Albinism
Chair: Grønskov K, Scalinci S

- Postnatal retinal development in albinism as compared to healthy controls – Irene Gottlob (20 min)
- Visual cortex in albinism - anatomy and function – Michael Hoffmann (20 min)
- Contribution of Electrofunctional Examinations in Albinism Diagnosis – Alma Patrizia Tormene (15 min)
- Molecular diagnostic approaches: Targeted sequencing, exome, array-CGH? - Benoit Arveiler (20 min)
- Discussion (15 min)
11:30-12:00 coffee break

12:00-13:30 Scientific Session III
European Diagnostic Experiences

Chair: Del Longo A, Taïeb A, Primignani P

- Clinical and molecular experience in Denmark – Karen Grønskov (20 min)
- Clinical and molecular experience in Italy – Roberta Terrana, Lucia Mauri (15 min)
- Albino Day in Madrid - Marta Cortón (15 min)
- Albino Day in Bordeaux – Fanny Maurice Picard (15 min)
- Discussion (20 min)

13:30-14:40 Lunch break

14:40-16:00 Scientific Session IV
Pathogenesis of Albinism

Chair: Montoliu L, Schiaffino MV,

- Understanding visual defects in albinism using mouse and cellular models - Alexandra Rebsam (20 min)
- Syndromic albinism disorders affecting membrane trafficking during melanosome formation- Michael Marks (20 min)
- Melanosomal ion channels critical for pigmentation – Elena Oancea (20 min)
- Discussion (20 min)

16:00-16.30 Coffee/Tea break

16:30-18:10 Scientific Session V:
Beyond Eyes

Chair: Arveiler B., Penco S.

- Oculocutaneous albinism in East and Southern Africa - Patricia Lund (20 min)
- Skin photoprotection issues in albinism - AlainTaïeb (20 min)
- Dermatological characterization in Italian albinos –Marco Antonio Negri (20 min)
- Audiological deficits in albinos: correlation with genetic/clinical phenotype – Giuseppe Mingoaia (20 min)
- Discussion (20 min)
Friday, 8 April 2016

8.30-10:00 Scientific Session VI
Technical Support and Therapeutic Frontiers

Chair: Käsmann-Kellner B, Del Longo A

- Visual management training in low vision patients - Flavia Fabiani (15 min)
- Pharmacological therapy – Brian Brooks (20 min)
- Defining safety and efficacy end-points for OCAI gene therapy-based clinical studies – Enrico Surace (15 min)
- New animal models of albinism generated with CRISPR tools – Lluis Montoliu (20 min)
- Discussion (20 min)

10.00 – 10.30 Final Scientific Discussion:

Chairs: Schiaffino MV, Marocchi A, Gottlob I, Montoliu L

- Discussion (30 min)

Plenary final session: scientists and associations together

- 10.30 -11:00 Summary of scientific presentations (Käsmann-Kellner B, Patrosso MC)
- 11:00 - 11.20 Summary of psycho pedagogical presentations (20 min)
- 11.20 -11.50 Summary of association’s presentations (30 min)
- 11.50- 12.00 Kid for kindness presentation (10 min)
- 12.00- 12.10 Dermatological Prevention presentation (10 min)
- 12.10-13.30 Global discussion/Presentation of leaflet/book on albinism for the general public/Proposal for the next meeting.